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PHOTOLETTER TO THE EDITOR

A neurocutaneous rarity: phacomatosis pigmentokeratotica

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Abstract

Phacomatosis pigmentokeratotica is characterized by the coexistence of nevus sebaceus, papular nevus spilus and associated neurologic abnormalities. We report a case of phacomatosis pigmentokeratotica in a 28-year-old male who presented with palmar-plantar dysesthesia and ipsilateral brain hemiatrophy. As a characteristic neuroimaging finding of the disorder, we found multiple hypointense lesions involving the ipsilateral hemisphere. (*J Dermatol Case Rep*. 2014; 8(2): 58-59)

Key words:

atrophy, bones, congenital abnormalities, dysesthesia, nevus sebaceous, neuropathy, nevus spilus, unilateral

Phacomatosis pigmentokeratotica (PPK) is a peculiar variant of Schimmelpenning syndrome being characterized by the coexistence of a sebaceous nevus and a speckled lentiginous nevus.[1,2] Central nervous system and neuromuscular disorders are an important feature of this rare disorder. Skeletal or other extracutaneous abnormalities may likewise occur.[1,2]

A 28-year-old man presented to the neurologic department with a history of dermatological lesions since birth. Lesions were characterized by nevus sebaceus and papular nevus spilus that predominantly involved his left side. They became more prominent at the age of five and progressively extended involving his scalp, face, arms, and trunk. The patient's dermatological diagnosis was PPK (Fig. 1A-C). At the age of 23, the patient complained of a painful and swollen left lower leg (Fig. 1D), and magnetic resonance imaging showed pronounced thickening of his left tibia and fibula as well as soft tissue edema (Fig. 2A). Two years later, the patient complained of left palmar and plantar dysesthesia which prompted a neurologic examination that revealed a mild proximal atrophy and weakness (4+ MRC muscle strength) of his left arm. Brain magnetic resonance imaging revealed left hypointense lesions in the cranium of the frontal, occipital, clivus and petrosal parts of the temporal bones (Fig. 2B) as well as congenital arachnoid cysts in the left parietal region (Fig. 2C) and the left temporal pole (Fig. 2D). In addition, brain MRI yielded hemiatrophy of the left cerebral hemisphere with widening of cortical sulci and ventricular asymmetry (Fig. 2B and C). His dysesthesia was not troublesome and therefore no treatment was performed.

PPK is characterized by the coexistence of nevus sebaceus and papular nevus spilus with associated neurologic abnormalities including mental deficiency, seizures, hemiparesis, hyperhidrosis, cutaneous dysesthesia, muscular weakness, and sensory or motor neuropathy.[2] Neuroimaging may show hemimegalencephaly, Dandy-Walker malformation, vascular dysplasia, agenesis of corpus callosum, cerebral heterotopia, and cortical agyria, microgyria or pachygyria.[1-3] In the present case we documented multiple hypointense lesions within one of the hemispheres. Skeletal and ocular defects are likewise noted. Lymphedema of one leg as noted in

our patient has previously been reported in this disorder.[3] The disorder can be taken as a particular variant of Schimmelpenning syndrome. Originally, PPK had been explained as a twin-spot phenomenon (didymosis).[2] Recently, however, molecular evidence was provided that the paired nevi originate from one single multipotent progenitor cell being heterozygous for an HRAS mutation.[4] Hence, the twin-spot theory can no longer be upheld and the disorder should be regarded as a "pseudodidymosis".[5] Remarkably, the spectrum of associated neurological anomalies appears to be different when either nevus occurs as an isolated cutaneous lesion. For example, segmental hyperhidrosis and dysesthesia are characteristic features of the nevus spilus component[2] and they appear to be absent in patients who show the nevus sebaceus component only.[3]

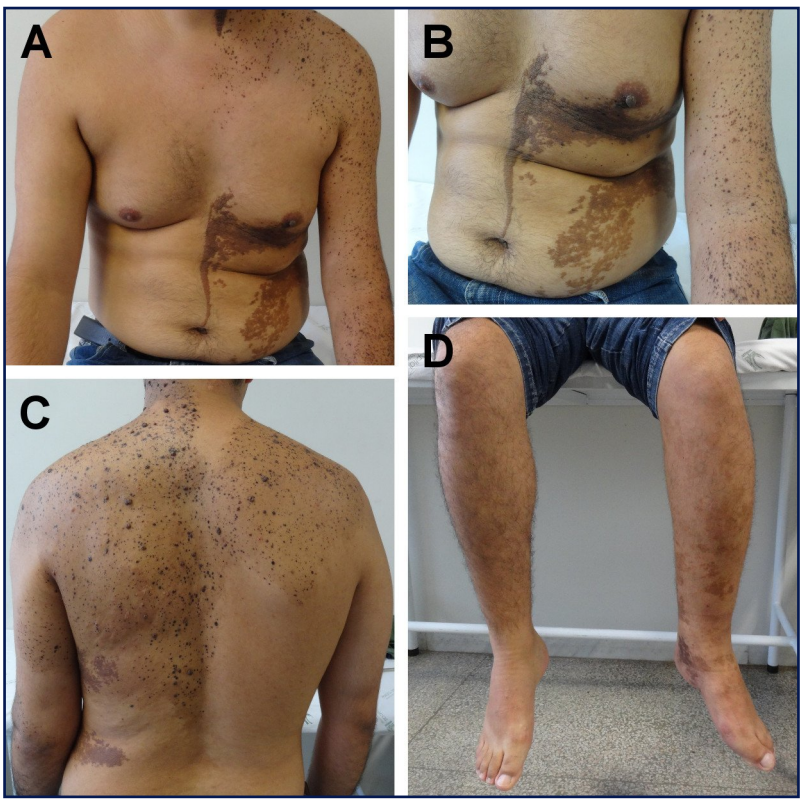

Figure 1

Phacomatosis pigmentokeratotica in a 28-year-old man. A-C: Congenital nevus sebaceus and papular nevus spilus that preponderantly involve his left side; D: Swelling of his left lower leg.

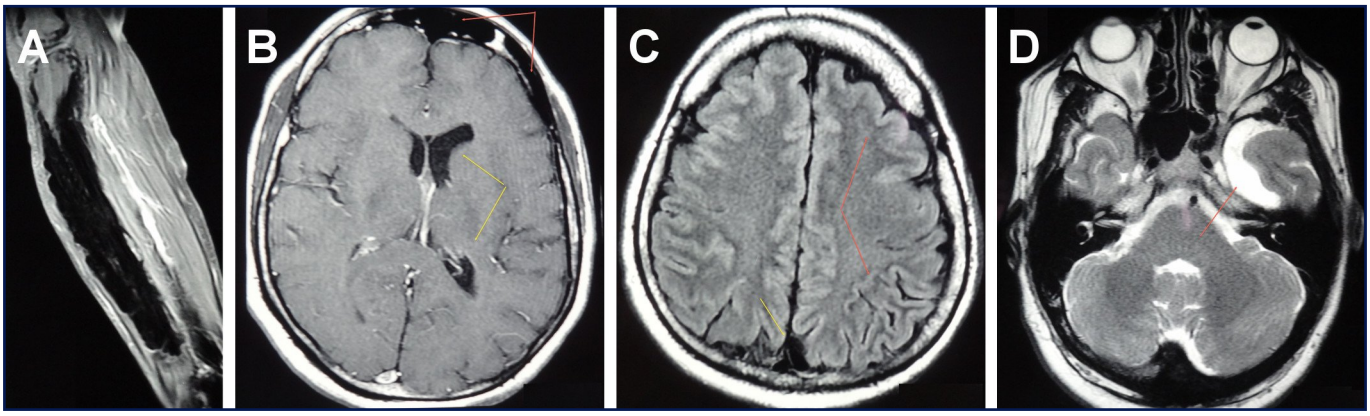

Figure 2

Radiological features of the patient. A: Thickening of his left tibia and fibula; B: T1-weighted brain MRI showing left hypointense lesions in the cranium of the frontal (red arrows), occipital, clivus and petrosal parts of the temporal regions, and ventricular asymmetry (yellow arrows); C: Congenital arachnoid cyst in the left parietal region (yellow arrow, T1-weighted brain MRI) and hemiatrophy of the left cerebral hemisphere with widening of cortical sulci (red arrows); D: Arachnoid cyst in the left temporal pole (red arrow, T2-weighted brain MRI).

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